

Note on expected internode distances for gene trees in species trees

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Abstract

In a recent paper on ‘Estimating Species Trees from Unrooted Gene Trees’ Liu and Yu observe that the distance matrix on the underlying taxon set, which is built up from expected internode distances on gene trees under the multispecies coalescent, is tree-like, and that the underlying additive tree has the same topology as the true species tree. Hence they suggest to use (observed) average internode distances on gene trees as an input for the neighbor joining algorithm to estimate the underlying species tree in a statistically consistent way. In this note we give a rigorous proof of their above mentioned observation.

1 Introduction

One of the possible reasons for discordance of a gene tree with an underlying species tree is the phenomenon of incomplete lineage sorting, which is described by the multispecies coalescent model. Many authors have addressed the problem of reconstructing the underlying species tree from a set of discordant gene trees, both from a theoretical perspective (e.g. Maddison [7], Allman et al. [1], and many others), as well as from an practical resp. algorithmic perspective (see e.g. Ewing et al. [3], Liu et al. [5], Than and Nakhleh [8], Kreidl [6]). Recently, Liu and Yu have published a paper [4] in which they propose to estimate the *expected number of internodes between any two taxa on gene trees* by averaging over the observed numbers of internodes (on the observed gene trees), for any pair of taxa. They note

Theorem 1 (Liu and Yu, [4]). *Under the multispecies coalescent model on any fixed species tree, the expected number of internodes between two taxa on gene trees determines a tree-like metric on the taxon set, and the underlying tree topology is identical with the topology of the species tree.*

Hence, they conclude, by applying the neighbor joining algorithm to the matrix of average internode distances (obtained from observed gene trees) is a statistically consistent way to estimate the true species tree. It is the goal of this note to give a rigorous and detailed proof of Liu and Yu’s theorem above.

We start by collecting, in Section 2, a few well-known facts on tree-like metrics, as well an easy reformulation of the *four-point condition* in terms of *weights of quartets* (for the terminology of weights of quartet trees see e.g. Sturmfels and Pachter [9]). Section 3, finally, contains the precise statement of Liu and Yu’s theorem together with its proof. The proof consists essentially in checking that the ‘weight-version’ of the four-point condition from Section 2 holds for the matrix of expected numbers of internodes.

I would like to thank Liang Liu for his interest in this modest note.

2 Preliminaries on tree-like metrics

In the following let T be a finite set and let $D : T \times T \rightarrow \mathbb{R}_{\geq 0}$ be a metric on T .

Definition 2. *The metric D satisfies the four-point condition if the maximum of the three numbers*

$$D(a, b) + D(c, d), \quad D(a, c) + D(b, d), \quad D(a, d) + D(b, c) \quad (2.1)$$

is attained at least twice, for every four-element subset $\{a, b, c, d\} \subset T$.

For every four taxon subset $\{a, b, c, d\} \subset T$ we define the *weight* of the quartet (ab, cd) , according to the exposition by Pachter and Sturmfels [9], to be the number

$$\begin{aligned} w(ab, cd) &:= w_D(ab, cd) = \\ &= D(a, c) + D(a, d) + D(b, c) + D(b, d) - 2D(a, b) - 2D(c, d). \end{aligned} \quad (2.2)$$

In the following we will sometimes consider weights with respect to different metrics D , which will be indicated by a lower index.

Definition 3. *The metric D is said to satisfy the weight condition if the minimum of the three numbers*

$$w_D(ab, cd), \quad w_D(ac, bd), \quad w_D(ad, bc) \quad (2.3)$$

is attained at least twice, for every four-element subset $\{a, b, c, d\} \subset T$.

Remark 4. (1) *The sum of the three numbers in the weight condition is always 0. Thus their minimum is strictly negative and their maximum is strictly positive as soon as not all three numbers vanish.*

(2) *If the metric D is tree-like and the underlying tree \mathcal{T} displays the quartet (ab, cd) , then $w(ab, cd) = 4x$ and $w(ac, bd) = w(ad, bc) = -2x$, where x is the distance between the paths connecting a and b , and c and d , respectively.*

Lemma 5. (1) *For a metric D the four-point condition and the weight condition are equivalent.*

(2) *The metric D is tree-like if and only if these conditions hold.*

(3) *If D is tree-like and \mathcal{T} is the underlying tree, then \mathcal{T} displays the quartet (ab, cd) if and only if $D(a, b) + D(c, d)$ is the minimum of the three numbers in the four-point condition if and only if $w(ab, cd)$ is the maximum of the three numbers in the weight condition.*

Proof. It is well known that if D satisfies the four point condition then D is tree-like, and moreover that the underlying tree displays the quartet (ab, cd) if and only if the minimum in the four point condition is attained at $D(a, b) + D(c, d)$ (for a proof see e.g. Pachter and Sturmfels [9]). From the remark above it follows that if D is tree-like with the underlying tree displaying the quartet (ab, cd) , then it satisfies the weight condition with maximum at $w(ab, cd)$. It remains to check that if D satisfies the weight condition with maximum at $w(ab, cd)$, then it satisfies the four-point condition with minimum at $D(a, b) + D(c, d)$. Thus assume that $w(ab, cd) > w(ac, bd) = w(ad, bc)$. Then

$$0 = w(ac, bd) - w(ad, bc) = 3(D(a, d) + D(b, c)) - 3(D(a, c) + D(b, d)).$$

Hence we obtain $D(a, d) + D(b, c) = D(a, c) + D(b, d)$. By plugging this into the definition of $w(ac, bd)$ we obtain

$$0 > w(ac, bd) = D(a, b) + D(c, d) - D(a, c) - D(b, d),$$

whence $D(a, b) + D(c, d) < D(a, d) + D(b, c) = D(a, c) + D(b, d)$, which completes the proof. \square

3 Expected internode distances on gene trees

We consider a taxon set $T = \{t_1, \dots, t_N\}$ containing N taxa, and a species tree S on T . Assume that for each taxon $t \in T$ we have sampled $n(t)$ copies of a given locus, denoted $L_{i,1}, \dots, L_{i,n(t_i)}$ for each $i = 1, \dots, N$. Let $\mathcal{L} = \cup_i \{L_{i,1}, \dots, L_{i,n(t_i)}\}$. We follow the convention to denote the elements of \mathcal{L} by capital letters, while leaves on the species tree S (i.e. the elements of T) are denoted by lower case letters.

For each rooted binary tree G on the leaf set \mathcal{L} Liu and Yu define in [4] the *internode distance* between leaves I and J to be the *number of nodes which lie on the path between I and J in G* (I and J are not counted). This number, which we denote $I_G(I, J)$, induces a metric on \mathcal{L} and thus a weight $W_G(IJ, KL) := w_{I_G}(IJ, KL)$ for each four element subset $\{I, J, K, L\} \subset \mathcal{L}$. Of course, for any G the metric I_G is tree-like with underlying tree G .

Definition 6. A coalescence pattern associated with the species tree S and the vector of multiplicities $(n(t_1), \dots, n(t_N))$ is a rooted tree G with leaf set \mathcal{L} together with a map

$$f : \text{Nodes}(G) \rightarrow \text{Nodes}(S)$$

with the following two properties: (1) For every $L_{i,j} \in \mathcal{L}$ we have $f(L_{i,j}) = t_i \in T$, and (2) For any two nodes $m, n \in \text{Nodes}(G)$, if n is a descendant of m in G , then $f(n)$ is a descendant of $f(m)$ in S . We denote coalescence patterns as pairs (G, f) in the sequel.

A coalescence pattern is basically the same as what Degnan and Salter [2] call a (valid) coalescent history. Each coalescence pattern (G, f) (associated with S and a multiplicity vector $v = (n(t_i))_i$) occurs with a certain probability $P(G, f)$ under the multispecies coalescent, which is calculated in

the case $n(t) = 1$ for all t by Degnan and Salter in loc. cit. This makes the set of coalescence patterns (for fixed S and v !) a probability space, and the internode distance $I_G(I, J)$ for each gene tree G between two leaves $I, J \in \mathcal{L}$ induces a random variable on this probability space, which we denote by $ID(I, J)$. By abuse of language we call this random variable also ‘internode distance’ between I and J . Similarly for the weight of a quartet (IJ, KL) for $I, J, K, L \in \mathcal{L}$: We denote the corresponding random variables $W(IJ, KL)$, for each quartet (IJ, KL) .

Thus the following numbers, associated with S and v , are well-defined (here and in the following we suppress the dependence on S and v in the notation, though we want to stress once more that all this requires choosing and fixing a multiplicity vector v !):

$$\begin{aligned} D(I, J) &= E(ID(I, J)) = \sum_{(G,f)} P(G, f) \cdot I_G(I, J), \\ E(W(IJ, KL)) &= \sum_{(G,f)} P(G, f) \cdot W_G(IJ, KL), \end{aligned} \tag{3.1}$$

the *expected internode distance* between two leaves $I, J \in \mathcal{L}$, and the expected weight of a quartet (IJ, KL) under the multispecies coalescent model.

Lemma 7. (1) *Since for each G the function I_G is a metric, so is $D = E(I)$. (2) Hence the weight function w_D is defined and satisfies $w_D(IJ, KL) = E(W(IJ, KL))$.*

Proof. Both claims are immediate consequences of linearity of expected values. \square

Finally, we note that the expression $D(I, J)$ does not really depend on the leaves I, J , but only on the taxa in T they belong to. Thus we have defined a metric

$$D(i, j) \in \mathbb{R}_{\geq 0}, \quad \text{for any two taxa } i, j \in T,$$

as well as a weight (depending on D)

$$w_D(ij, kl) \in \mathbb{R}, \quad \text{for any four taxa } i, j, k, l \in T.$$

Combining Equation (3.1) with Lemma 7 we obtain that we may calculate the weight $w_D(ij, kl)$ for four taxa $i, j, k, l \in T$ as

$$w_D(ij, kl) = \sum_{(G, f)} P(G, f) \cdot W_G(IJ, KL), \quad (3.2)$$

where (G, f) runs through all possible coalescence patterns, and where $I \in \mathcal{L}$ is any locus corresponding to $i \in T$, J any locus corresponding to $j \in T$ and so forth.

Theorem 8 (Liu and Yu, [4], Theorem A1). *The metric $D = E(I)$ is tree-like, and the underlying additive tree has the same topology as the true species tree S .*

For the proof we introduce a little piece of notation: For any rooted tree \mathcal{T} and a finite subset of leaves l_1, \dots, l_k we denote by $M_{\mathcal{T}}(l_1, \dots, l_k)$ the *most recent common ancestor* of l_1, \dots, l_k in \mathcal{T} .

Proof. By Lemma 5 it suffices to check that, if the species tree S displays the quartet (ab, cd) , then the following holds:

$$w_D(ab, cd) > w_D(ac, bd) = w_D(ad, bc).$$

This is relatively easy to check using equation (3.2). We thus assume that S displays the quartet (ab, cd) , and we consider gene lineages A, B, C, D sampled from the respective taxa. We have to distinguish two cases, namely: (1) The (rooted) subtree S' of S with leaf set $\{a, b, c, d\}$ has the shape of a caterpillar tree, and (2) S' has the balanced shape. In case (1) we assume without loss of generality that S' has the topology $((a, b), c), d$, while in the second S' must have the topology $((a, b), (c, d))$. S' has the balanced shape. In case (1) we assume without loss of generality that S' has the topology $((a, b), c), d$, while in the second case S' must have the topology $((a, b), (c, d))$.

We now partition the set of coalescence patterns (G, f) into two disjoint subsets X and Y : In case (1) we define

$$\begin{aligned} X &= \{(G, f) \mid f(M_G(A, B)) \text{ is ancestral to } M_S(a, b, c)\}, \\ Y &= \{(G, f) \mid (G, f) \notin X\}. \end{aligned} \quad (3.3)$$

Note that $(G, f) \in P$ then means that the lineages A and B coalesce below the point where the populations c merges with the population ancestral to a and b . In case (2) we set

$$\begin{aligned} X &= \{(G, f) \mid f(M_G(A, B)) \text{ and } f(M_G(C, D)) \\ &\quad \text{are both ancestral to } M_S(a, b, c, d)\}, \\ Y &= \{(G, f) \mid (G, f) \notin Q\}. \end{aligned} \quad (3.4)$$

Consider the case of a coalescence pattern $(G_1, f_1) \in X$. Then the lineages of A , B and C on G_1 enter the population above $M_S(a, b, c)$ separately. Hence, by permuting the lineages A , B and C we obtain coalescence patterns $(G_2, f_2), (G_3, f_3) \in X$ such that $P(G_1, f_1) = P(G_2, f_2) = P(G_3, f_3)$, and such that, after possibly renumbering of the coalescence patterns, G_1 displays (AB, CD) , G_2 displays (AC, BD) and G_3 displays (AD, BC) , and such that

$$\begin{aligned} -2W_{G_1}(AC, BD) &= -2W_{G_1}(AD, BC) = W_{G_1}(AB, CD) = x, \\ -2W_{G_2}(AB, CD) &= -2W_{G_2}(AD, BC) = W_{G_2}(AC, BD) = x, \\ -2W_{G_3}(AB, CD) &= -2W_{G_3}(AC, BD) = W_{G_3}(AD, BC) = x, \end{aligned} \quad (3.5)$$

where x is the number of nodes on the path connecting the path between A and B , and C and D , respectively, in G_1 .

On the other hand, if $(G, f) \in Y$, then G necessarily displays the quartet (AB, CD) . Hence for such G we have

$$\begin{aligned} W_G(AB, CD) &> 0, \quad \text{while} \\ W_G(AC, BD) &= W_G(AD, BC) = -\frac{1}{2}W_G(AB, CD). \end{aligned} \quad (3.6)$$

Now recall equation (3.2) and write

$$\begin{aligned} w_D(ij, kl) &= \sum_{(G, f)} P(G, f) \cdot W_G(IJ, KL) = \\ &= \sum_{(G, f) \in X} P(G, f) \cdot W_G(IJ, KL) + \sum_{(G, f) \in Y} P(G, f) \cdot W_G(IJ, KL) \end{aligned}$$

From Equation (3.5) we see that in the expressions $w_D(ab, cd)$, $w_D(ac, bd)$ and $w_D(ad, bc)$ the sum over the $(G, f) \in X$ vanishes, and equation (3.6)

further implies that

$$\begin{aligned}
w_D(ab, cd) &= \sum_{(G,f) \in Y} P(G, f) \cdot W_G(AB, CD) > 0, \quad \text{while} \\
w_D(ac, bd) = w_D(ad, bc) &= \sum_{(G,f) \in Y} P(G, f) \cdot -\frac{1}{2}W_G(AB, CD) = \\
&= -\frac{1}{2}w_D(ab, cd)
\end{aligned}
\tag{3.7}$$

This shows that D satisfies the weight condition, with the maximum attained for the quartet (ab, cd) . Invoking Lemma 5 completes the proof. \square

References

- [1] Elizabeth S. Allman, James H. Degnan, and John A. Rhodes. *Identifying the rooted species tree from the distribution of unrooted gene trees under the coalescent*. J. Math. Biol. **62** no. 6 (2011), 833–862.
- [2] J. H. Degnan and L. A. Salter. *Gene tree distributions under the coalescent process*. Evolution (2005), 59:24–37.
- [3] Gregory B. Ewing, Ingo Ebersberger, Heiko A. Schmidt, Arndt von Haeseler. *Rooted triple consensus and anomalous gene trees*. BMC Evol Biol (2008), 8:118.
- [4] Liang Liu and Lili Yu. *Estimating species trees from unrooted gene trees*. Syst. Biol. **60** no. 5 (2011) 661–667.
- [5] Liang Liu, Lili Yu and Scott V. Edwards. *A maximum pseudo-likelihood approach for estimating species trees under the coalescent model*. BMC Evolutionary Biology (2010), 10:302.
- [6] Martin Kreidl. *Estimating Species Trees from Quartet Gene Tree Distributions under the Coalescent Model*. Preprint (2011), arXiv:1108.1628v1.
- [7] Wayne Maddison. *Gene trees in species trees*. Syst. Biol. (1997), 46(3):523–536.
- [8] C. Than and L. Nakhleh, *Species tree inference by minimizing deep coalescences*. PLoS Computational Biology (2009), 5(9): e1000501.

- [9] Lior Pachter and Bernd Sturmfels. *Algebraic Statistics for Computational Biology*. Cambridge University Press (2005).